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Original article

A podoscopic and descriptive study of foot deformities in patients with Down syndrome

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ABSTRACT

Introduction: Subjects with Down syndrome (DS) are known to be affected by various foot deformities. Despite the fact that some of these deformities have been reported in the literature, a more comprehensive description would be of benefit. The aim of this study is to investigate the prevalence of known foot deformities in patients with DS and of other previously non-described foot anomalies in this population.
Hypothesis: Subjects with DS have an increased prevalence of foot deformities compared to control subjects.
Methods: Fifty-five subjects with DS (age: 14.6 ± 7.4 years) had undergone podiatric clinical and podoscopic examinations to study their main foot deformities and their footprints, respectively. The results of these examinations were compared to those of an age-matched asymptomatic control group of fifty-three subjects (age: 13.4 ± 11.2 years).
Results: Significantly more prevalent foot deformities were found in the DS group: hallux valgus (36.4%), syndactyly between the 2nd and 3rd toes (9.1%), grade II pes planus (39.1%) and grade III pes planus (30%). Moreover, joint laxity (43.6%) was significantly more prevalent in the DS group. Furthermore, the presence of an increased space between the 1st and 2nd toes in patients with DS and its prevalence (73.6%) were described for the first time. A multivariate analysis revealed a significant relationship between the presence of joint laxity and flatfoot in only the control but not the DS group. Other foot deformities were found to be consistently more frequent in the DS population but not significantly higher than the control group.
Discussion: Although subjects with DS had significantly greater joint laxity and BMI compared to the control group, neither of these factors was found to be related to the increased prevalence of flatfoot in DS patients.
Level of evidence: IV–retrospective study.

1. Introduction

With an incidence of 1/660 live births, Down syndrome (DS) also known as trisomy 21 is the most frequent chromosomal abnormality [1]. This syndrome is associated with a multitude of anomalies. While the management of these patients generally focuses on the detection and treatment of life-threatening defects such as cardiac, respiratory and neoplastic diseases [1], there is a growing need regarding treatment and management of orthopedic anomalies due to their impact on quality of life of these patients.

Normal foot morphology is essential for normal gait and some affections of the foot may be inter-related to more proximal lower limb anomalies. While most authors have focused on the major orthopedic deformities in patients with DS such as scoliosis [2], atlanto-axial instability [3], hip subluxation, slipped capital femoral epiphysis, Legg-Calve-Perthes syndrome and patellar instability [1,2,4], a few authors have investigated less debilitating orthopedic anomalies of the foot such as flatfoot [5,6], hallux valgus [4,5,7], syndactyly [5,7] and clinodactyly [7]. Despite the reported high prevalence of foot anomalies in DS populations, the current literature is far from being exhaustive and does not include investigations on other foot deformities such as hallux varus, overriding fifth toe (quintus varus supraductus), short metatarsals and metatarsus adductus.

Moreover, a control group originating from the same ethnic origin as the DS group is essential in order to conclude significantly

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higher prevalences of anomalies in patients with DS, since it has been shown that the phenotype of patients with DS is, at least in part, influenced by their parents' genotypes [8]. None of the extensive descriptive studies of foot anomalies inpatients with DS have compared their reported prevalences to age-matched control groups [6,7].

Furthermore, not all patients with DS present the same podiatric and podoscopic abnormalities. Thus, this variety of anomalies might be related to other etiologic factors and therefore some abnormalities of the feet could be related to each other and to other factors such as joint laxity, gender or weight. Only one study has investigated a few clinically significant correlations between anomalies found in patients with DS [7]. More correlation studies would be beneficial in understanding the potential causal or coincidental associations between the observed anomalies.

Thus, the primary aim of this study is to describe foot deformities in patients with DS compared to an age-matched control group. The secondary objective of this study is to explore relationships between these anomalies and possible etiological factors. Our hypothesis was that subjects with DS have an increased prevalence of a large number of foot anomalies and that this prevalence could be, at least partly, explained by the increased weight and joint laxity of these subjects.

2. Materials and methods

This is a cross-sectional observational IRB approved study of fifty-five patients with clinically [9] and genetically diagnosed DS (36 males, 19 females, mean age: 14.6 years, SD: 7.4) who have no musculoskeletal complaints and no previous history of orthopedic surgery. Fifty-three asymptomatic subjects (27 males, 26 females, mean age: 13.5 years, SD: 11.2), which were accompanying family members at orthopedic consultations, with no previous orthopedic history were enrolled in the study as an age-matched control group. An informed consent form was signed by all the subjects and patients when applicable, or their legal guardians. Age, gender, height, weight and body mass index (BMI) were recorded for each subject. Furthermore, joint laxity was defined as follows: when the wrist is in extreme flexion, the passive abduction of the thumb brings it to a position at least parallel to the anterior surface of the forearm (and touching the forearm in extreme cases), as previously described by Carter and Wilkinson [3,10]. Podiatric and podometric examinations were carried out by 2 senior orthopedic residents, in the presence of an experienced orthopedic surgeon.

2.1. Podiatric clinical examination

A thorough podiatric clinical examination was performed for the two groups in an effort to identify any of the following foot deformities: hallux valgus, hallux varus, overriding fifth toe, metatarsus adductus, widened space between first and second toes (Fig. 1), short metatarsals, syndactyly and/or clinodactyly [11].

Widened space between the first and second toes was defined as having parallel toes with no contact between them, even when there is no frank hallux varus. Hallux valgus was defined as the presence of a lateral bony prominence over the metatarsophalangeal joint of the hallux associated with a lateral deviation of the hallux. Hallux varus was defined as a medial deviation of the hallux with or without an associated widened space between the first and second toes. These deformities were only evaluated by clinical examination and neither control subjects nor subjects with DS underwent foot radiographs.

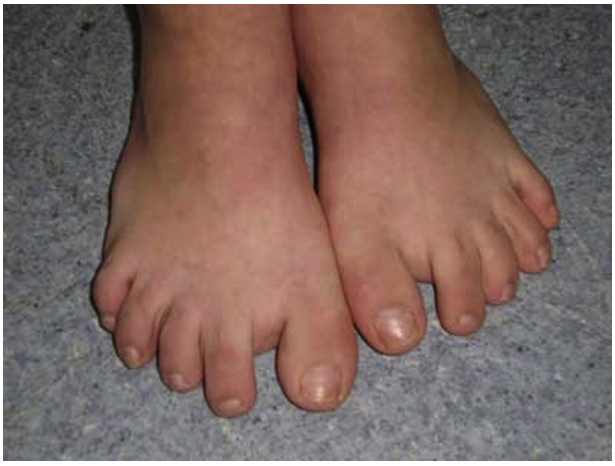

Fig. 1. Increased space between first and second toes.

2.2. Podometry

Footprints were assessed on a podoscope (Podometer, Pedihealth Ltd), and were classified qualitatively into: normal footprint, pes cavus, pes planus grades I, II and III [12] (Fig. 2).

2.3. Statistical study

All quantitative variables were tested for normality using the Shapiro-Wilk test. Mean comparison tests were used in order to compare the various characteristics of the two groups. Anthropometric measurements of control and DS groups were compared using the Mann–Whitney U test. Significant differences in deformity prevalence were evaluated between the control and DS groups using Pearson's chi-squared test and Fisher's exact test. In order to control for age, gender, anthropometric characteristics and joint laxity when comparing the prevalence of foot deformities between the two groups, an ordinal regression (with the flatfoot grades as the dependent variable) and a series of binary logistic regressions (with the rest of the foot deformities as dependent variables) were computed. The effect of subject characteristics on the prevalence of deformities was evaluated by the odds ratio (and its 95% confidence interval). The significance level was set at $P < 0.05$. Statistical analysis was performed using SPSS Statistics version 20.0 (IBM Corporation, New York, USA) and Xlstat version 2015.3.1 (Addinsoft, Paris, France).

3. Results

3.1. Anthropometric measurements

Anthropometric measurements of the DS and control subjects are reported in Table 1. The average weight did not significantly

Table 1
Comparison of anthropometric parameters between DS and control groups. The medians and interquartile ranges (in brackets) are reported for age, weight, height and BMI.

	DS group	Control group	*P*-value
Number of subjects	55	53	
Ratio of males to females	36:19	27:26	0.109
Age (years)	14 (10–17)	11 (6–15)	0.550
Weight (kg)	45.5 (31.9–55.5)	41.0 (28.0–55.0)	0.282
Height (m)	1.37 (1.28–1.44)	1.50 (1.18–1.60)	0.016*
BMI (kg/m^2)	23.2 (18.4–26.8)	20.5 (16.2–23.4)	0.041*

*Significant differences.

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